sub I 1 #1

21. (Twice Amended) A kit for the *in vitro* detection of a truncation, a deletion or a mutation in the survival motor neuron gene, comprising:

a set of primers wherein said primers are contained within the sequence of nucleotides 921 to 1469 of SEQ ID No: 12;

reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

sub I 2 #2

30. (Twice Amended) A method for detecting a truncation, a deletion or a mutation in the Survival Motor Neuron gene, said method comprising:

(a) extracting DNA from a patient sample;

(b) amplifying said DNA with primers, wherein said primers are contained in the sequence of nucleotides 921 to 1469 of SEQ ID No: 12;

(c) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP) analysis; and

(d) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene, wherein the presence of said truncation, deletion or mutation is indicative of a Survival Motor Neuron disorder.

31. (Twice Amended) The method of Claim 30, wherein said detection of a truncation, deletion or mutation in the Survival Motor Neuron gene is indicative of a Spinal Muscular Atrophy.

#3

32. (Once Amended) The method of Claim 30, wherein step (c) is replaced with a step of digestion with a *Bsr-1* enzyme.

#4

36. (Twice Amended) A method for detecting Arthrogryposis Multiplex Congenita (AMC), said method comprising :

(a) extracting DNA from a patient sample;

(b) amplifying said DNA via a polymerase chain reaction (PCR) using unlabeled primers from exon 7 or exon 8 of the Survival Motor Neuron (SMN) gene of SEQ ID No:22;

(c) subjecting said amplified DNA to a Single Stranded Conformation Polymorphism (SSCP) analysis; and

(d) detecting the presence or absence of Arthrogryposis Multiplex Congenita.

40. (Twice Amended) A method of detecting the presence in a human patient of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy, comprising:

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID No: 22) in a biological sample derived from the patient, and

comparing said exon 7 to the corresponding exon from nucleotide position 340 to nucleotide position 401 of SEQ ID No:13, or exon 8 to the corresponding exon from nucleotide position 846 to nucleotide position 1408 of SEQ ID No:13, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy in said patient.

43. (Once amended) The method of claim 40, wherein said analyzing includes amplifying all or part of the T-BCD541 gene.

50. (Twice Amended) A method of confirming a clinical diagnosis of Arthrogryposis Multiplex Congenita in a patient, comprising

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID No : 22) in a biological sample derived from the patient, and

comparing said exon 7 to the corresponding exon from nucleotide position 340 to nucleotide position 401 of SEQ ID No:13, or exon 8 to the

corresponding exon from nucleotide position 846 to nucleotide position 1408 of SEQ ID No:13, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Arthrogryposis Multiplex Congenita in said patient.

53. (Once amended) A kit for the *in vitro* detection of a defect in the Survival Motor Neuron gene, wherein said kit comprises a probe which comprises at least 9 nucleotides within a sequence of SEQ ID No: 22 or hybridizes under stringent conditions with a sequence of SEQ ID Nos: 1, 2, 10-13, or 22.

54. (Once amended) A method of identifying the presence or absence of a mutation in the Survival Motor Neuron (SMN) gene in a subject, comprising

(a) isolating a nucleic acid from the subject;

(b) subjecting the nucleic acid to digestion by a restriction endonuclease, wherein restriction fragments resulting from said digestion of a mutated SMN gene differ from those obtained from a T-BCD541 gene of SEQ ID No:22; and

(c) identifying the presence or absence of a mutation in the SMN gene in the subject.

57. (Once amended) The method of claim 56, wherein said polymerase chain reaction is performed with a set of primers which are contained in the sequence comprising nucleotides 921 to 1469 of SEQ ID No: 12, or which comprise a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57.

58. (Once amended) A method of identifying the presence of Spinal Muscular Atrophy (SMA) in a subject, said method comprising:

(a) isolating a nucleic acid from a subject; and

sub E7

(b) identifying a mutation in a T-BCD541 gene (SEQ ID No: 22);

wherein the presence of a mutation in the T-BCD541 gene is indicative of the presence of SMA in said subject.

#9 Cont

59. (Once amended) The method of claim 58, wherein the mutation is a deletion in the T-BCD541 gene (SEQ ID No: 22).

60. (Once amended) The method of claim 59, wherein the deletion comprises a deletion of the entire T-BCD541 gene (SEQ ID No: 22).

sub I8

64. (Once amended) A kit for the *in vitro* detection of a defect in the survival motor neuron gene, comprising:

a set of primers wherein said primers comprise a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57;

reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

#10

65. (Once amended) A method for detecting a defect in the Survival Motor Neuron gene, said method comprising :

(a) extracting DNA from a patient sample;

(b) amplifying said DNA with primers, wherein said primers comprise a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57;

(c) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP); and

(d) detecting the presence or absence of said defect in the Survival Motor Neuron gene, wherein the presence of said defect is indicative of a Survival Motor Neuron disorder.